

## Familial labial pits: A report of three cases

Alok Dubey, MDS

تعتبر التجاويف الشفوية العائلية إصابة وراثية تؤدي إلى تشكل عيوب تطورية حول الجزء الأوسط لحواف الشفتين العلوية والسفلية أو منطقة الصوار الشفوية. تظهر هذه العيوب بشكل متناظر حول حواف الخط الأوسط للشفة في المستوى الجبهي على شكل فتحات بيضوية مغلقة النهايات. تعرض هذه المقالة وصفا لحالة طفلة بعمر 12 سنة لديها تجويف في الشفة إضافة لشقيقها ووالدتها.

Familial labial pit is an autosomal dominant trait resulting in developmental defects involving the paramedial portion of the vermilion of the lower and upper lip or the labial commissure area. This deformity of the vermilion border of the lower lip constantly appears symmetrically to the midline in a frontal direction with blind ending ducts and oval openings. Cases of lip pit in a 12-year-old girl, her brother and mother are presented.

### INTRODUCTION

Normal morphogenesis of oro-facial tissues includes a complete, step-by-step sequence of cell migration and of interaction between groups of cells whereas mechanisms responsible for dysmorphogenesis are still unknown. Lips are frequent target structures for these malformations, a variety of which are congenital labial fistula.<sup>1</sup>

In contrast to the upper cleft lip and/or cleft palate, congenital bilateral fistulas of the lower lip form a new, distinct syndrome explained by an arrested development leading to cross sulci resulting from a pathological cleaving process. In most cases a striking inheritance pattern with an autosomal dominant trait is associated with this disease.<sup>2</sup>

Congenital lip pits (fistula labii inferioris congenital) may occur alone or in combination with oral clefts and other dysontogenetic malformations such as Van der Woude syndrome and Kabuki syndrome.<sup>3</sup> Clinically, they appear as small pits on the lip with or without excretion. They may appear as bumps on an infant's lip, changing to depressions as the child grows older. Excretion as seen in these lip pits is associated with salivary glands.<sup>3</sup>

Lip pits may have variable clinical aspects, representing several degrees of gene expression. Pits are usually bilateral but may be unilateral or centrally placed on vermilion border of lower lip.<sup>2</sup> There may be a single pit in the center of lip, two pits bilaterally to the lip midline, or one pit on either the right or left side. Affected individuals may have maxillary hypodontia or missing premolars.<sup>4</sup>

### CASE REPORT

A twelve-year-old female patient, of Indian nationality, presented for regular dental check up along with her mother. On clinical examination, bilateral punctate depressions were seen on her lower lip, about 0.5 cm from the midline (Fig. 1). No secretion was either visible or expressible in the lip pits. Clinical examination of her mother also revealed similar lip pits on her lower lip. The girl's brother reported to our OPD, the following day. His clinical examination also revealed isolated lip pits without secretion, with no other oral abnormality (Fig. 2). Both children had apparently normal intelligence with no cleft lip or palate.

Surgical excision of the lip pits was advised, to improve the appearance of lips. However, the patient refused surgical intervention as she had no discomfort associated with the pits neither did she agree to any surgery for cosmetic reasons.

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Specialist Pedodontist

Regional Dental Centre, King Khalid Hospital

P.O. Box 1120, Najran, KSA

E-mail: dentistalok@yahoo.co.in



**Fig. 1.** Clinical picture of the child and her mother showing distinct paramedian lower lip pits.



**Fig. 2.** Clinical picture of the child's brother showing similar isolated pits on the lower lip.

## DISCUSSION

Congenital pits of the lower lip are rare malformations, inherited as an autosomal dominant with a penetrance of approximately 80 percent.<sup>5</sup> Interstitial deletion of chromosome 1q [(1q32----41)] has been linked to congenital lip pits.<sup>6</sup>

The condition usually appears in several members and in several generations of the same family. Both sexes are affected almost equally. The depth

of the pits sometimes varies among the affected members of the same family this being interpreted as variable expressivity of the trait.<sup>7</sup>

Surgical excision of sinus tract (lip pits) is often performed either to improve appearance of lip pits or reduce mucous drainage.<sup>8</sup> The sinus tract consists of stratified non-keratinized squamous epithelium and lamina propria of dense connective tissue.<sup>9</sup>

The recognition of familial lip pits has relevance for genetic counseling. Physical examination of relatives, close examination of family photos or interview of older relatives may be necessary to identify minimally affected family members. However, because of variable expressivity of phenotype, the potential effects on unborn children are difficult to predict and high resolution ultrasound and fetal echo cardiography may be of some use in characterizing the severity of phenotype.

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